

Supplemental Figure S3. Identification of genes

implicated in development and manifestations of hundreds
physiological and pathological phenotypes and autosomal
inheritance in Modern Humans using the Enrichr
bioinformatics platform (see Methods for details).

Human Phenotype Ontology: 8,405 genes

Autosomal dominant inheritance (HP:0000006)

Downslanted palpebral fissures (HP:0000494)

Cryptorchidism (HP:0000028)

Autosomal recessive inheritance (HP:0000007)

Hypertelorism (HP:0000316)

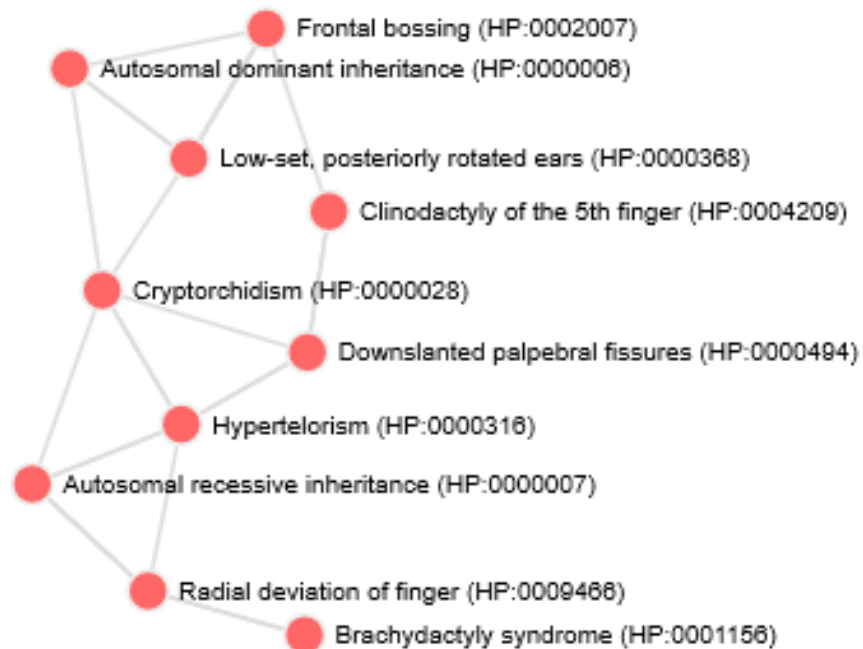
Radial deviation of finger (HP:0009466)

Brachydactyly syndrome (HP:0001156)

Clinodactyly of the 5th finger (HP:0004209)

Frontal bossing (HP:0002007)

Low-set, posteriorly rotated ears (HP:0000368)

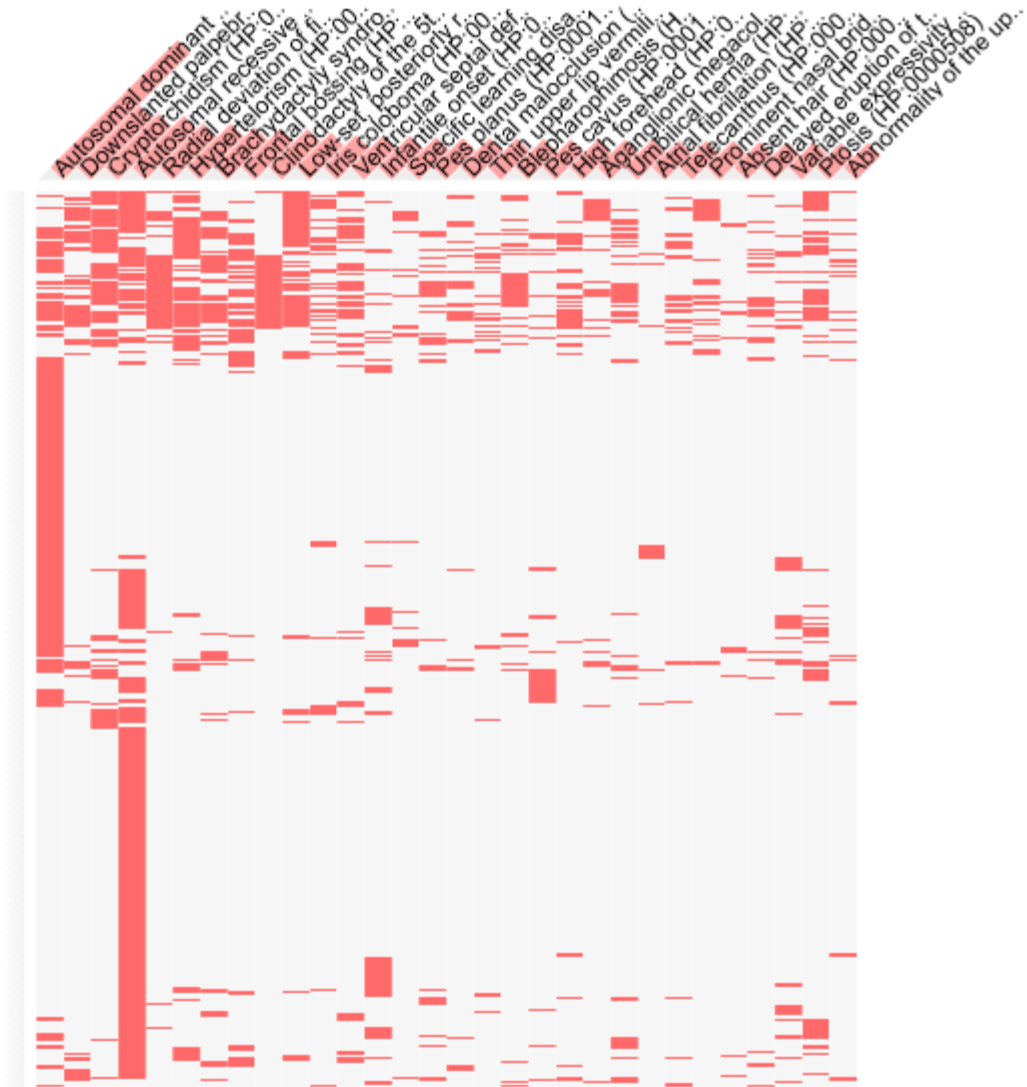


Human Phenotype Ontology: 8,405 genes



Enriched Terms

Input Genes



Human Phenotype Ontology (8,405 genes): Top 35 of 298 significant records

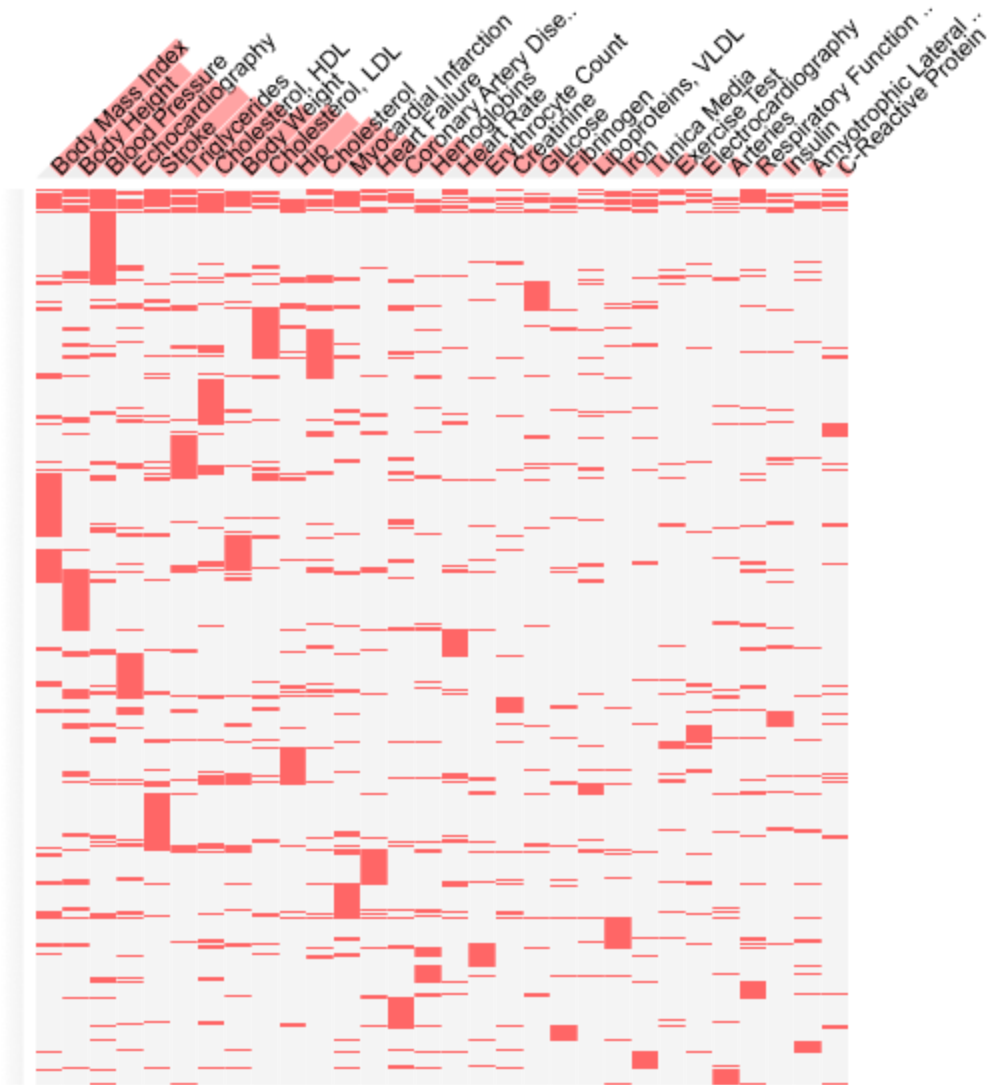
Term	Overlap	P-value	Adjusted P-value
Autosomal dominant inheritance (HP:0000006)	645/1134	2.85E-25	5.07E-22
Downslanted palpebral fissures (HP:0000494)	127/188	1.17E-12	1.04E-09
Cryptorchidism (HP:0000028)	222/379	4.46E-11	2.64E-08
Autosomal recessive inheritance (HP:0000007)	849/1722	1.14E-10	5.07E-08
Radial deviation of finger (HP:0009466)	140/229	3.75E-09	1.11E-06
Hypertelorism (HP:0000316)	190/328	3.76E-09	1.11E-06
Brachydactyly syndrome (HP:0001156)	119/192	1.77E-08	4.5E-06
Frontal bossing (HP:0002007)	129/213	3.38E-08	7.51E-06
Clinodactyly of the 5th finger (HP:0004209)	118/192	4.03E-08	7.96E-06
Low-set, posteriorly rotated ears (HP:0000368)	158/276	2.14E-07	3.8E-05
Iris coloboma (HP:0000612)	72/110	5.77E-07	9.34E-05
Ventricular septal defect (HP:0001629)	106/176	7.99E-07	0.000113
Infantile onset (HP:0003593)	145/254	8.28E-07	0.000113
Specific learning disability (HP:0001328)	36/47	1.49E-06	0.000189
Pes planus (HP:0001763)	68/105	2.09E-06	0.000246
Dental malocclusion (HP:0000689)	55/81	2.21E-06	0.000246
Thin upper lip vermilion (HP:0000219)	38/51	2.5E-06	0.000261
Blepharophimosis (HP:0000581)	67/104	3.26E-06	0.000323
Pes cavus (HP:0001761)	80/129	3.55E-06	0.000323
High forehead (HP:0000348)	69/108	3.63E-06	0.000323
Aganglionic megacolon (HP:0002251)	63/97	4.26E-06	0.000361
Umbilical hernia (HP:0001537)	91/151	4.5E-06	0.000364
Atrial fibrillation (HP:0005110)	26/32	6.52E-06	0.000505
Telecanthus (HP:0000506)	55/83	6.96E-06	0.000516
Prominent nasal bridge (HP:0000426)	64/100	7.43E-06	0.000529
Absent hair (HP:0002298)	18/20	1.14E-05	0.000781
Delayed eruption of teeth (HP:0000684)	56/86	1.28E-05	0.000841
Variable expressivity (HP:0003828)	86/144	1.34E-05	0.00085
Ptosis (HP:0000508)	180/338	1.79E-05	0.001074
Abnormality of the upper arm (HP:0001454)	29/38	1.81E-05	0.001074
Progressive disorder (HP:0003676)	86/145	1.94E-05	0.001103
Depressed nasal bridge (HP:0005280)	127/228	1.99E-05	0.001103
Sporadic (HP:0003745)	42/61	2.05E-05	0.001103
Left ventricular hypertrophy (HP:0001712)	31/42	2.93E-05	0.0015
Postaxial hand polydactyly (HP:0001162)	53/82	2.95E-05	0.0015

Database of Human Genotypes and Phenotypes (dbGaP): 8,405 genes



Enriched Terms

Input Genes



Database of Human Genotypes and Phenotypes (dbGaP): 8,405 genes
Top 20 of 136 significant records

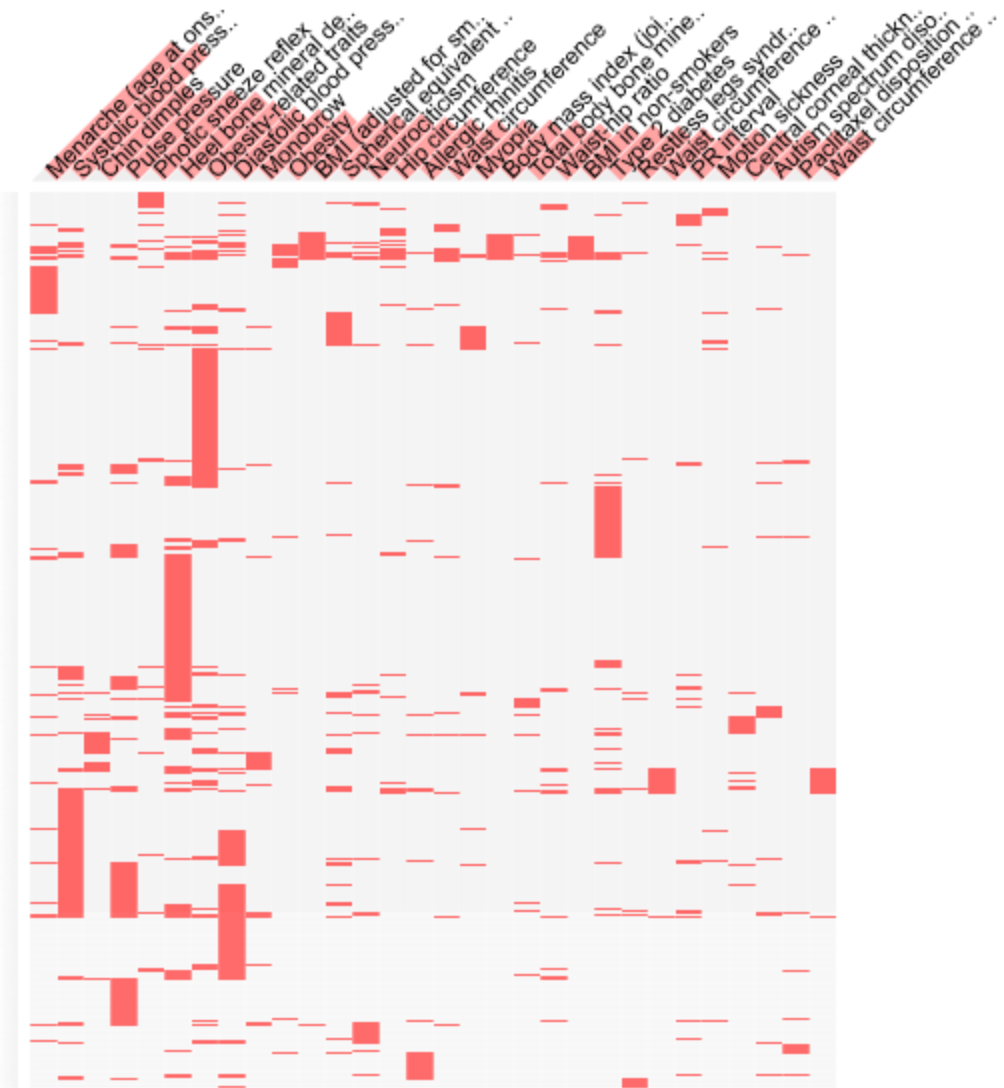
Term	Overlap	P-value	Adjusted P-value
Body Mass Index	313/437	1.02E-36	3.46E-34
Body Height	276/385	1.32E-32	2.23E-30
Blood Pressure	310/454	3.19E-30	3.59E-28
Echocardiography	204/273	2.47E-28	2.08E-26
Stroke	211/289	6.02E-27	4.07E-25
Triglycerides	180/244	4.77E-24	2.68E-22
Cholesterol, HDL	242/357	3.57E-23	1.72E-21
Body Weight	161/216	1.91E-22	8.05E-21
Cholesterol, LDL	210/304	7.78E-22	2.92E-20
Hip	151/202	2.39E-21	8.06E-20
Cholesterol	183/268	2.28E-18	7.02E-17
Myocardial Infarction	157/229	3.32E-16	9.36E-15
Coronary Artery Disease	140/205	2.16E-14	5.31E-13
Heart Failure	130/187	2.2E-14	5.31E-13
Hemoglobins	113/157	2.38E-14	5.37E-13
Erythrocyte Count	87/115	2.09E-13	4.41E-12
Heart Rate	110/155	2.47E-13	4.86E-12
Creatinine	73/92	2.59E-13	4.86E-12
Fibrinogen	69/86	4.29E-13	7.05E-12
Lipoproteins, VLDL	69/86	4.29E-13	7.05E-12

GWAS Catalog 2019: 8,405 genes



Enriched Terms

Input Genes



GWAS Catalog 2019 (8,405 genes): Top 40 of 241 significant records

Term	Overlap	P-value	Adjusted P-value
Menarche (age at onset)	154/212	1.12E-19	1.4E-16
Systolic blood pressure	389/657	1.62E-19	1.4E-16
Chin dimples	62/74	1.51E-13	8.68E-11
Pulse pressure	321/567	9.66E-13	4.16E-10
Photic sneeze reflex	55/65	1.6E-12	5.51E-10
Heel bone mineral density	478/898	3.09E-12	8.88E-10
Obesity-related traits	427/804	6.98E-11	1.72E-08
Diastolic blood pressure	348/646	4.87E-10	1.05E-07
Monobrow	58/76	1.19E-09	2.28E-07
Obesity	45/55	1.61E-09	2.78E-07
Height	288/527	2.32E-09	3.63E-07
Atrial fibrillation	146/240	2.83E-09	3.78E-07
BMI (adjusted for smoking behaviour)	58/77	2.85E-09	3.78E-07
Spherical equivalent or myopia (age of diagnosis)	120/191	4.87E-09	5.99E-07
Neuroticism	69/97	5.87E-09	6.74E-07
Hip circumference	52/69	1.83E-08	1.97E-06
Diverticular disease	100/156	2.04E-08	2.06E-06
Allergic rhinitis	77/114	3.13E-08	3E-06
Waist circumference	55/75	3.67E-08	3.33E-06
Myopia	52/70	4.2E-08	3.62E-06
Body mass index (joint analysis main effects and smoking interaction)	56/77	4.5E-08	3.69E-06
Hand grip strength	99/156	5.05E-08	3.96E-06
Total body bone mineral density	31/36	5.77E-08	4.32E-06
Waist-hip ratio	44/58	1.65E-07	1.14E-05
BMI in non-smokers	44/58	1.65E-07	1.14E-05
Type 2 diabetes	217/397	2.08E-07	1.38E-05
Restless legs syndrome	32/39	3.48E-07	2.22E-05
Blond vs. brown/black hair color	98/160	6.9E-07	4.25E-05
Waist circumference adjusted for BMI (adjusted for smoking behaviour)	56/81	7.21E-07	4.29E-05
Amyotrophic lateral sclerosis (sporadic)	109/182	8.3E-07	4.77E-05
PR interval	48/67	8.6E-07	4.78E-05
Motion sickness	27/32	1.01E-06	5.45E-05
Male-pattern baldness	143/251	1.15E-06	6E-05
Central corneal thickness	47/66	1.48E-06	7.14E-05
Paclitaxel disposition in epithelial ovarian cancer	36/47	1.49E-06	7.14E-05
Autism spectrum disorder, ADHD, bipolar disorder, MDD, and schizophrenia (combined)	36/47	1.49E-06	7.14E-05
Rosacea symptom severity	87/141	1.84E-06	8.57E-05
Waist circumference adjusted for BMI (joint analysis main effects and smoking interaction)	54/79	1.98E-06	8.99E-05
Waist circumference adjusted for body mass index	80/128	2.29E-06	0.000101
Glaucoma (primary open-angle)	52/76	2.92E-06	0.000124